# SLC39A14 gene

solute carrier family 39 member 14

#### **Normal Function**

The *SLC39A14* gene provides instructions for making a protein that transports the element manganese across cell membranes. Manganese is important for many cellular functions, but large amounts are toxic, particularly to brain cells. The SLC39A14 protein is found in the membranes surrounding several types of cells, as well as in the membranes of structures within these cells. The protein is thought to transport excess manganese from the blood into liver cells so that it can be removed from the body through bile. Bile is a substance produced by the liver that is important for digestion and the removal of waste products.

The SLC39A14 protein may also transport other elements, including zinc, iron, and cadmium, across cell membranes. The importance of this transport in the body is not well understood.

# **Health Conditions Related to Genetic Changes**

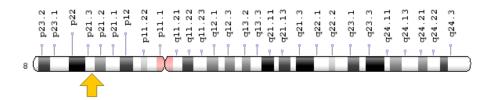
## Hypermanganesemia with dystonia

At least five *SLC39A14* gene mutations have been found to cause hypermanganesemia with dystonia 2, a condition that begins in early childhood and is characterized by high levels of manganese in the blood and brain (hypermanganesemia), involuntary tensing of the muscles (dystonia), and other movement problems. These mutations impair the transport of manganese into liver cells. As a result, the element cannot be removed from the body through bile. The excess manganese builds up in the blood and subsequently in brain cells, particularly cells in a region of the brain that helps control movement. High levels of manganese damage these cells, causing neurological problems that make controlling movement difficult.

## **Chromosomal Location**

Cytogenetic Location: 8p21.3, which is the short (p) arm of chromosome 8 at position 21.3

Molecular Location: base pairs 22,367,278 to 22,434,129 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- cig19
- HMNDYT2
- KIAA0062
- LIV-1 subfamily of ZIP zinc transporter 4
- LZT-Hs4
- NET34
- solute carrier family 39 (metal ion transporter), member 14
- solute carrier family 39 (zinc transporter), member 14
- ZIP14
- Zrt-, Irt-like protein 14
- zrt- and Irt-like protein 14

#### **Additional Information & Resources**

#### **Educational Resources**

- Agency for Toxic Substances and Disease Registry: Manganese https://www.atsdr.cdc.gov/substances/toxsubstance.asp?toxid=23
- Oregon State University Linus Pauling Institute: Manganese https://lpi.oregonstate.edu/mic/minerals/manganese

#### Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SLC39A14%5BTIAB%5D%29+OR+%28solute+carrier+family+39+member+14%5BTIAB%5D%29\*29+OR+%28%28LIV-1+subfamily+of+ZIP+zinc+transporter+4%5BTIAB%5D%29+OR+%28ZIP14%5BTIAB%5D%29+OR+%28Zrt-,+Irt-like+protein+14%5BTIAB%5D%29+OR+%28y+0R+%28solute+carrier+family+39++,+member+14%5BTIAB%5D%29+OR+%28zrt-+and+Irt-like+protein+14%5BTIAB%5D%29\*29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

## Catalog of Genes and Diseases from OMIM

 SOLUTE CARRIER FAMILY 39 (ZINC TRANSPORTER), MEMBER 14 http://omim.org/entry/608736

#### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/GC\_SLC39A14.html
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=SLC39A14%5Bgene%5D
- HGNC Gene Symbol Report https://www.genenames.org/data/gene-symbol-report/#!/hgnc\_id/HGNC:20858
- Monarch Initiative https://monarchinitiative.org/gene/NCBIGene:23516
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/23516
- UniProt https://www.uniprot.org/uniprot/Q15043

## **Sources for This Summary**

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